

Case 2

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Clinical Abstract:

The patient was a 13 year old girl born after a term gestation to a 21 year old mother with a history of poor weight gain and little fetal activity during pregnancy. She weighed 5 lb. 2 oz. and was 17 in. long at birth. Her development was characterized as quiet with feeding difficulties. From the age of 10 months on she had numerous recurrent fevers, urinary tract infections and episodes of tonsillitis. Extensive neurologic and psychologic evaluations at 3 years and 5 years of age disclosed petit mal episodes occurring up to 5 times per day in a child with short stature, ataxic gait and psychomotor development equivalent to an 18 to 21 month old. She sat with prodding a 1 year, walked alone at 2 1/2 years of age and spoke only a few words by age 5, although she appeared to comprehend and asked for things with gestures. Her family history was remarkable for parents of short stature, two maternal great uncles with mental slowness and short stature and a normal younger sibling. She attended a special school for several years. From the age of 8 until her death at age 13 she became increasingly dependent on her parents, with an increase in unsteadiness, numerous falls, regression of her speech and developed an increasingly irritable personality. She had a cataract removed at age 10 1/2. In the 2 months prior to her death she could walk only a few steps without falling and had several hospital admissions for periods of lethargy after such falls. In the months prior to her death, she lost bladder and bowel control, developed a resting tremor and exhibited continuous jitteriness and irritability. On her last hospital admission, she had a 3 day period of lethargy after a fall. CT scan showed subdural hemorrhage over the left cerebral hemisphere. She deteriorated steadily into coma and death.

Autopsy findings: She was a cachectic small child, only 38 inches tall. Flexion contractures of the extremities were present and focal calcification of the heart, adrenals and kidney were found. She had evidence of bilateral subdural hemorrhages of varying ages with distortion of the underlying cerebral hemispheres. A single section is available for review.

Material Submitted: One H & E Stained Section

Points for discussion: Diagnosis